Summer 8-2-2022

Unmasking Beta Thalassemia

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Introduction

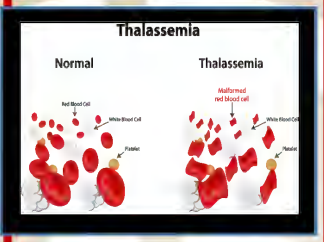
As a practitioner, it is important to understand the disease prevalence in the community in which care is provided. For example, rural Ohio is home to a large population of Amish. Amish have a higher prevalence of disease based on the lifestyle that they live. Having an inquiry as to why things are the way they are is an initial introduction to forming a research question. Evidence based practice, leads the practitioner to information that will help guide them to conclusions and answers in the management of improving or understanding patient's situations.

Beta-Thalassemia

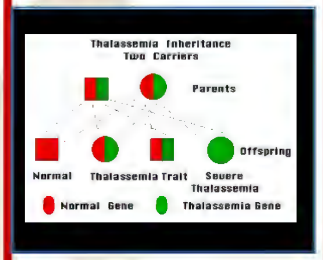
Autosomal Recessive

- One mutated gene from both parents.
- Parents carriers NOT affected

The importance of understanding a specific disease process is to help identify treatment options that fit best with the patient's values and wishes. (Khan et al., 2018)



Credit: joshya/Shutterstock.com



(Dorr, 2003)

The WHY Behind the Pain

Seeing my husband struggle prior to his diagnosis of beta-thalassemia was the most difficult situation I have gone through. Being a nurse, family and friends often come asking for advice on their minor ailments or lab results interpretation. However, in my husband's situation we were battling the unknown. It started with what we thought was a back and groin strain from working out. This ended him in the bed after being discharged from the emergency room with muscle relaxers and heating pads.

Reoccurrence of this type of injury occurred and had me worried so imaging was performed, and this is where the nightmare started The physicians interpreted his MRI scan as him having cancer. My world stopped spinning and at that moment in time I knew what the doctors were saying and where their minds were going based on the conversation. The frustrating thing was, is that I was now in the role of the patient's spouse and emotions were high.. One oncologist later and we were cleared on the cancer diagnosis and back to square one. Blood work was the next work up which revealed my husband indeed had the beta thalassemia diagnosis. This explained the long nights of pain, in the back and groin and issues with his vision (which we later found out can cause vessel occlusion or calcium deposits), and the emotional and mental distress of having a lifelong disease. I chose this topic to gain more understanding about the disease and treatment options available so that I can be an advocate for my husband's health.

The Various Signs and Symptoms 3 Main Forms:

Beta-thalassemia major:

- Diagnosed at early infancy/toddler
- Require RBC transfusion
- Growth restriction s
- Nutrition deficiency
- Pale (Orega, 2017)

Beta-thalassemia intermedia:

- Diagnosed in middle adulthood
- Pale
- Bone frailty
- Blood clots
- Pseudoxanthoma elasticum- calcium deposits in skin, eyes, and blood vessels (Orega, 2017)

Beta-thalassemia trait

- Asymptomatic
- May have mild anemia (Orega, 2017)

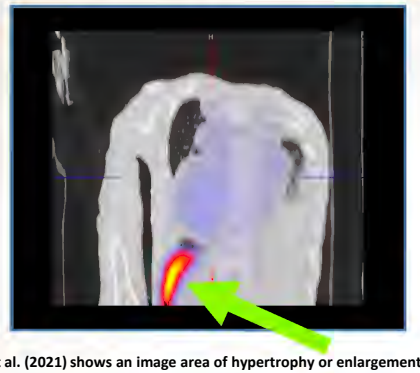
Pathophysiology of Beta Thalassemia

Primary

1. Alpha-globulin accumulation sticks to erythropoietin
2. Inclusion bodies formed
3. Inclusion bodies stick to membrane
4. Cause oxidative damage
5. Premature apoptosis of precursor RBCs
6. Accumulation of precursor RBCs in the bone marrow (Orega, 2017)

Secondary

1. Hypertrophy of erythroid bone marrow
2. Increase in bone sites causing deformity
3. Leading to fractures
4. Masses in the bone (Orega, 2017)



Kopparthy et al. (2021) shows an image area of hypertrophy or enlargement of the liver.

Significance of Pathophysiology

Genetic counseling/prenatal screening

Incidence is higher in the Mediterranean population due to uptick in trend of consanguinity (marrying of descendants of the same ancestor)

Newborn screening is sent to the Ohio Board of of Health to identify any congenital issues or serious diseases. Also includes a hearing screen. (Ohio Department of Health, 2022).

Implications in Nursing

The practitioner can prescribe **medications** that will help with symptoms:

- Folic Acid due to deficiency in the bone marrow
- Regular lab level checks to check for necessity of blood administration
- Anticoagulation for blood clot prevention
- Possible iron chelators to help with the increase in iron stores

Possible **Surgical** Interventions

- Splenectomy
- Colectomy due to the removal of the spleen

ONE IDENTIFIED CURITIVE PROCEDURE

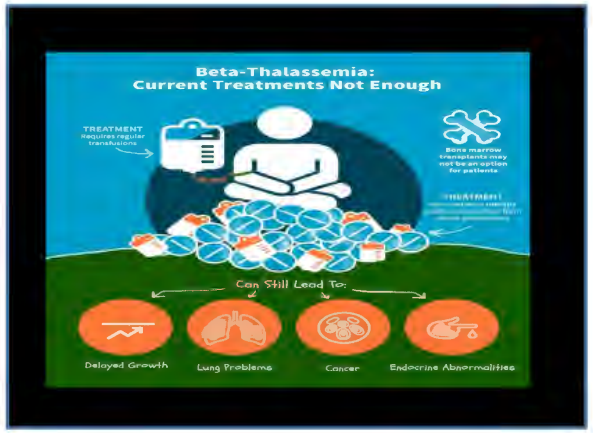
Bone marrow transplant, however, must be from an identical sibling. (Origa, 2017)

Gene Addition Strategy

Trials are underway that correct the mutated gene and add in the functional gene. Testing is done using lentivectors which are a type of retrovirus that has a strong ability to destroy the membrane of the nucleus of the normal cell. (Dong & Rivella, 2017).

Vitamin D Usage

Thalassemia causes deficiency of supplements in the bone, one of which is calcium. For calcium absorption to be optimal, correlation of vitamin D also plays a role. There are test being conducted to confirm if supplementation with vitamin D improves symptoms of patients with thalassemia. (Manolopoulos et al., 2021)



(Rajan, 2018)

Conclusion

Living with a genetic disease can create stress for the patient who is already suffering from uncomfortable symptoms depending on the severity rating of the beta thallesemia they have. The goal of the practitioner should be to help aide and guide the patient in the best treatment options based on their environment, lifestyle, and financial situation. During the planning phase, inclusion of patient support systems are very important currently. Research guided by a clinical question allows for best practices and better patient outcomes.

References

Additional Resources

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